

# 受賞講演 / Award Lecture

## 受賞講演 1 受賞講演 / Award Lecture 1 Award Lecture

日時：11月17日(金) 15:25～17:20 第1会場 (B1F メインホール)  
座長：松原 洋一 (国立成育医療研究センター研究所)  
Date: Friday, Nov.17 15:25～17:20 Room1 (Main Hall, B1F)  
Chair: Yoichi Matsubara (National Center for Child Health and Development)

### 学会賞 2016 小児先天性疾患の臨床・分子遺伝学的研究

#### Clinical and molecular genetic in congenital disorders

○緒方 勤 (Tsutomu Ogata)

浜松医科大学小児科

(Department of Pediatrics, Hamamatsu University School of Medicine)

### 学会賞 2017 分子細胞遺伝学的アプローチによる癌と遺伝性疾患のゲノム・エピゲノム解析研究

#### Genomic and epigenomic analysis of cancer and genetic disorders by molecular cytogenetic approach

○稲澤 譲治 (Johji Inazawa)

東京医科歯科大学難治疾患研究所 分子細胞遺伝

(Medical Research Institute, Tokyo Medical and Dental University)

### 貢献賞 2016 Bone dysplasia families

○西村 玄 (Gen Nishimura)

埼玉医大病院、難病センター

(Intractable Disease Center, Saitama Medical University)

### 貢献賞 2017 わが国の臨床遺伝医療のあゆみ

#### Clinical Genetics in Japan

○福嶋 義光 (Yoshimitsu Fukushima)

信州大学 医学部

(Shinshu University School of Medicine)

### 奨励賞 16-1 染色体工学技術を用いた新規ダウン症候群モデル細胞・動物の作製と解析

#### Generation of novel Down's syndrome model mice and cells via chromosome engineering technology and the characterization

○香月 康宏 (Yasuhiro Kazuki)

鳥取大学大学院医学系研究科 染色体工学研究センター

(Chromosome Engineering Research Center (CERC) and Graduate School of Medical Science, Tottori University)

### 奨励賞 16-2 骨・関節疾患の感受性遺伝子の研究

#### Studies on Susceptibility Genes for Bone and Joint Diseases

○中島 正宏 (Masahiro Nakajima)

理化学研究所統合生命医科学研究センター骨関節疾患研究チーム

(Laboratory for Bone and Joint Diseases, Center for Integrative Medical Science, RIKEN)

### 奨励賞 16-3 次世代シーケンスデータの解析手法の開発と、日本人及び肝臓癌の全ゲノム解析

#### Comprehensive variant analysis of a Japanese individual and liver cancer genomes

○藤本 明洋<sup>1,2</sup> (Akihiro Fujimoto)

1 京都大学大学院 医学研究科 創薬医学講座

(Department of Drug Discovery Medicine, Graduate School of Medicine, Kyoto University)

2 理化学研究所 統合生命医科学研究センター

(RIKEN, Center for Integrative Medical Sciences, Yokohama, Japan)

奨励賞 17-1

### 新規先天性疾患MIRAGE症候群の発見と疾患概念確立

#### Discovery of MIRAGE syndrome

○鳴海 覚志 (Satoshi Narumi)

国立成育医療研究センター 研究所 分子内分泌研究部

(Department of Molecular Endocrinology, National Research Institute for Child Health and Development)

奨励賞 17-2

### ゲノム解析を通じた希少自己免疫性疾患の病態解明と治療応用

#### Genetic analyses of rare autoimmune diseases for clarification of pathophysiology and development of new treatment

○寺尾 知可史 (Chikashi Terao)

理化学研究所 統合生命医科学研究センター 統計解析研究チーム

(Laboratory for Statistical Analysis, RIKEN Center for Integrative Medical Sciences)

奨励賞 17-3

### 神経筋疾患(単一遺伝子病からrare variant関連疾患まで)の原因解明

#### Genetic study on neuromuscular disorders

○宮武 聡子 (Satoko Miyatake)

横浜市立大学遺伝子診療部

(Clinical Genetics Department, Yokohama City University Hospital)

## 受賞講演 2 国際 JHG 賞受賞講演

### Award Lecture 2 The Journal of Human Genetics Young Scientist Award Lectures

日時：11月17日(金) 17:20～18:20 第1会場 (B1F メインホール)

座長：松本 直通(横浜市立大学大学院医学研究科遺伝学)

Date: Friday, Nov.17 17:20～18:20 Room1 (Main Hall, B1F)

Chair: Naomichi Matsumoto (Department of Human Genetics, Yokohama City University Graduate School of Medicine)

JHG16-1

### Comprehensive DNA methylation analysis of peripheral blood cells derived from patients with first-episode schizophrenia

○Masaki Nishioka<sup>1,2</sup>、Miki Bundo<sup>1,3</sup>、Kiyoto Kasai<sup>2</sup>、Kazuya Iwamoto<sup>1,3</sup>

1 Department of Molecular Psychiatry, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan

2 Department of Neuropsychiatry, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan

3 Department of Molecular Brain Science, Graduate School of Medical Sciences, Kumamoto University, Kumamoto, Japan

JHG16-2

### Mutation spectrum and genotype-phenotype correlation of hearing loss patients caused by SLC26A4 mutations in the Japanese: a large cohort study

○Maiko Miyagawa、Shin-ya Nishio、Shin-ichi Usami

Department of Otorhinolaryngology, Shinshu University School of Medicine

JHG16-3

### The Indonesian Archipelago: An Ancient Genetic Highway Linking Asia and the Pacific

○Meryanne K. Tumonggor<sup>1,2</sup>、Tatiana Karafet<sup>3</sup>、Brian Hallmark<sup>3</sup>、J. Stephen Lansing<sup>1,4</sup>、Herawati Sudoyo<sup>2</sup>、Michael Hammer<sup>3</sup>、Murray Cox<sup>5</sup>

1 School of Anthropology, University of Arizona

2 Eijkman Institute for Molecular Biology

3 Arizona Research Laboratories, Division of Biotechnology, University of Arizona

4 Santa Fe Institute

5 Institute of Fundamental Sciences, Massey University

JHG17-1

### Downregulation of the microRNA-1/133a cluster enhances cancer cell migration and invasion in lung-squamous cell carcinoma via regulation of Coronin1C

○Hiroko Mataka<sup>1</sup>、Takeshi Chiyomaru<sup>2</sup>、Keiko Mizuno<sup>1</sup>、Hiromasa Inoue<sup>1</sup>、Naohiko Seki<sup>3</sup>

1 Department of Pulmonary Medicine, Graduate School of Medical and Dental Sciences, Kagoshima University

2 Department of Urology, National Hospital Organization Kagoshima Medical Center

3 Department of Functional Genomics, Graduate School of Medicine, Chiba University

JHG17-2

## Chaperone therapy for Krabbe disease: potential for patients with late-onset GALC mutations

○ Mohammad Arif Hossain<sup>1</sup>, Katsumi Higaki<sup>2</sup>, Hitoshi Sakuraba<sup>3</sup>, Yoshiyuki Suzuki<sup>4</sup>, Keiichi Ozono<sup>1</sup>, Yoshikatsu Eto<sup>5</sup>, Norio Sakai<sup>1</sup>

- 1 Department of Pediatrics, Osaka University Graduate School of Medicine
- 2 Division of Functional Genomics, Tottori University
- 3 Department of Clinical Genetics, Meiji Pharmaceutical University
- 4 Tokyo Metropolitan Institute of Medical Science
- 5 Advanced Clinical Research Center, Institute of Neurological Disorders

JHG17-3

## The first story is about connecting the dots - Bridging the gap between genes, brain and behavior

○ Sara Mascheretti<sup>1</sup>, Vittoria Trezzi<sup>1</sup>, Valentina Riva<sup>1</sup>, Denis Peruzzo<sup>2</sup>, Andrea Nordio<sup>2</sup>, Roberto Giorda<sup>3</sup>, Cecilia Marino<sup>4</sup>, Ginette Dionne<sup>5</sup>, Andrea Facoetti<sup>1,6</sup>, Simone Gori<sup>1,7</sup>, Filippo Arrigoni<sup>2</sup>

- 1 Child Psychopathology Unit Scientific Institute, IRCCS Eugenio Medea
- 2 Neuroimaging Lab - Scientific Institute, IRCCS Eugenio Medea
- 3 Molecular Biology Lab, Scientific Institute, IRCCS Eugenio Medea
- 4 Centre for Addiction and Mental Health, University of Toronto
- 5 School of Psychology, Laval University
- 6 Developmental Cognitive Neuroscience Lab, Department of General Psychology, University of Padua
- 7 Department of Human and Social Sciences, University of Bergamo